Higher risk of death among MEN1 patients with mutations in the JunD interacting domain: a Groupe d'etude des Tumeurs Endocrines (GTE) cohort study.

Titre	Higher risk of death among MEN1 patients with mutations in the JunD interacting domain: a Groupe d'etude des Tumeurs Endocrines (GTE) cohort study.
Type de publication	Article de revue
Auteur	Thevenon, Julien [1], Bourredjem, Abderrahmane [2], Faivre, Laurence [3], Cardot-Bauters, Catherine [4], Calender, Alain [5], Murat, Arnaud [6], Giraud, Sophie [7], Niccoli, Patricia [8], Odou, Marie-Françoise [9], Borson-Chazot, Françoise [10], Barlier, Anne [11], Lombard-Bohas, Catherine [12], Clauser, Eric [13], Tabarin, Antoine [14], Parfait, Béatrice [15], Chabre, Olivier [16], Castermans, Emilie [17], Beckers, Albert [18], Ruszniewski, Philippe [19], Le Bras, Morgane [20], Delemer, Brigitte [21], Bouchard, Philippe [22], Guilhem, Isabelle [23], Rohmer, Vincent [24], Goichot, Bernard [25], Caron, Philippe [26], Baudin, Eric [27], Chanson, Philippe [28], Groussin, Lionel [29], Boullay, Hélène Du [30], Weryha, Georges [31], Lecomte, Pierre [32], Penfornis, Alfred [33], Bihan, Hélène [34], Archambeaud, Françoise [35], Kerlan, Véronique [36], Duron, Françoise [37], Kuhn, Jean-Marc [38], Vergès, Bruno [39], Rodier, Michel [40], Renard, Michel [41], Sadoul, Jean-Louis [42], Binquet, Christine [43], Goudet, Pierre [44]
Pays	Royaume-Uni
Editeur	Oxford University Press (OUP)
Ville	Oxford
Type	Article scientifique dans une revue à comité de lecture
Année	2013
Langue	Anglais
Date	2013 May 15
Numéro	10
Pagination	1940-1948
Volume	22
Titre de la revue	Human Molecular Genetics
ISSN	1460-2083
Mots-clés	Family [45], Female [46], Follow-Up Studies [47], Humans [48], Male [49], Multiple Endocrine Neoplasia Type 1 [50], Mutation [51], Protein Structure, Tertiary [52], Proto-Oncogene Proteins [53], Proto-Oncogene Proteins c-jun [54], Risk Factors [55]

Résumé en anglais	Multiple endocrine neoplasia syndrome type 1 (MEN1), which is secondary to mutation of the MEN1 gene, is a rare autosomal-dominant disease that predisposes mutation carriers to endocrine tumors. Although genotype-phenotype studies have so far failed to identify any statistical correlations, some families harbor recurrent tumor patterns. The function of MENIN is unclear, but has been described through the discovery of its interacting partners. Mutations in the interacting domains of MENIN functional partners have been shown to directly alter its regulation abilities. We report on a cohort of MEN1 patients from the Groupe d'étude des Tumeurs Endocrines. Patients with a molecular diagnosis and a clinical follow-up, totaling 262 families and 806 patients, were included. Associations between mutation type, location or interacting factors of the MENIN protein and death as well as the occurrence of MEN1-related tumors were tested using a frailty Cox model to adjust for potential heterogeneity across families. Accounting for the heterogeneity across families, the overall risk of death was significantly higher when mutations affected the JunD interacting domain (adjusted HR = 1.88: 95%-CI = 1.15-3.07). Patients had a higher risk of death from cancers of the MEN1 spectrum (HR = 2.34; 95%-CI = 1.23-4.43). This genotype-phenotype correlation study confirmed the lack of direct genotype-phenotype correlations. However, patients with mutations affecting the JunD interacting domain had a higher risk of death secondary to a MEN1 tumor and should thus be considered for surgical indications, genetic counseling and follow-up.
URL de la notice	http://okina.univ-angers.fr/publications/ua10557 [56]
DOI	10.1093/hmg/ddt039 [57]
Titre abrégé	Hum. Mol. Genet.
Identifiant (ID) PubMed	23376981 [58]

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